Isolated Congenital Diaphragmatic Hernia in the Third Trimester: A Case Report and Literature Review

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SUMMARY

Objective: To report a rare congenital anomaly, a right diaphragmatic hernia, in a near-term baby.

Case Report: A 40-year-old female, gravida 3, para 2, had undergone regular prenatal care in our department since the early second trimester. She underwent amniocentesis at 16 weeks of gestation. The result showed normal 46,XY. Fetal growth was appropriate throughout the pregnancy. A small heart with marked left-side deviation was noted in the third trimester. The heart rate was less than 25% of normal. A homogenous mass with centralized vessels was noted in the fetus's right chest. The baby showed respiratory distress immediately after delivery. Imaging studies after birth proved there was a right diaphragmatic hernia with severe pulmonary hypertension and poor lung function.

Conclusion: Right congenital diaphragmatic hernia is rare. A prenatal diagnosis is difficult to make in the second trimester. Prognosis is greatly influenced by the associated abnormalities. [*Taiwanese J Obstet Gynecol* 2006;45 (1):83–85]

Key Words: diaphragmatic hernia, pulmonary hypoplasia

Introduction

Human lung development is generally divided into five stages: embryonic, pseudoglandular, canalicular, saccular, and alveolar. The lung can be visualized on ultrasound throughout the second and third trimesters. It appears as a homogenously echogenic, glandular tissue surrounding the heart. The diaphragm can also be visualized on longitudinal scans and is more apparent during the second trimester [1].

Fetal chest masses, unlike those in adults, are rarely malignant. They are harmful because of secondary physiologic and anatomic alterations such as lung hypoplasia, an important sequela. Lung masses rarely cause life-threatening lung hypoplasia, but defects such as congenital diaphragmatic hernia (CDH) do. Herein, we present a case of isolated right CDH in the third trimester that was not associated with any other structural abnormalities or aneuploidy.

Case Report

A 40-year-old female, gravida 3, para 2, had undergone regular prenatal care in our department since the early second trimester. She underwent amniocentesis at 16 weeks of gestation due to her advanced maternal age. Karyotyping showed a normal 46,XY fetus. Level II ultrasound at 20 weeks showed no obvious structural anomalies. Fetal growth was normal until 36 weeks of gestation. A small heart with marked left-side deviation was noted at 34 weeks. The chest heart rate was less than 25% of the normal ratio, and a homogenous mass with centralized vessels and a gall bladder-like bubble was noted in the right thoracic cavity. Neither stomach bubbles nor liver parenchyma could be seen (Figure 1). The normal hypoechoic diaphragmatic muscle could

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Figure 1. Chest heart rate is less than 25%, and a homogenous mass with centralized vessels and a gall bladder-like bubble is noted in the right thoracic cavity.

not be detected on longitudinal scan. Results of a non-stress test were within normal limits. Under the impression of an intrathoracic tumor or diaphragmatic herniation with marked lung and heart compression, prompt delivery was suggested (by cesarean section due to a previous cesarean section). A baby boy was delivered in the vertex position by hand and fundus pressure, with a body weight of 2,600 g and Apgar scores of 5 and 6 at 1 and 5 minutes, respectively. Respiratory distress occurred immediately after clamping of the umbilical cord. The neonate was immediately intubated by the pediatrician and sent to the intensive care unit for further evaluation. Imaging studies after birth showed a right diaphragmatic hernia with severe pulmonary hypertension and poor lung function (Figure 2).

The baby underwent surgical intervention on the third day and was discharged uneventfully 2 weeks later. The images after the operation showed a satisfactory lung condition except for mild pulmonary hemorrhage, which was probably caused by the surgery (Figure 3). The baby's condition and respiratory function improved on the 6th postoperative day, and imaging showed no progression of the collapsed right lower lung.

The growth chart and lung function of this baby were satisfactory 1 month after the operation, and seemed no different from other normal babies.

Discussion

Common intrathoracic masses in fetuses include CDH, congenital cystic adenomatoid malformation, bronchopulmonary sequestration, and bronchogenic cyst. All of these may result in pulmonary hypoplasia

Figure 2. Images show an opacity over the right lower lung field and liver with bowels herniated upward into the right thoracic cage. Left deviation of the mediastinum and heart are also noted.

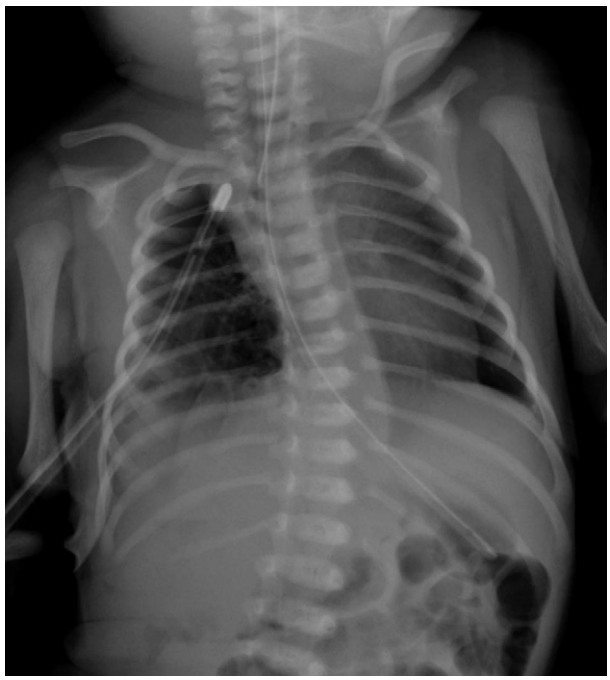

Figure 3. First postoperative day (3rd day after birth): a clear lung field but some pulmonary hemorrhage over the right lung parenchyma.

secondary to the mass effect at the time of onset. The diagnosis and prediction of pulmonary hypoplasia remain questionable at present [2]. Herein, we present a rare case of right CDH that occurred in the third trimester.

CDH is herniation of the abdominal viscera into the fetal chest because of a congenital defect in the diaphragm. It is the most common abnormality of the diaphragm, occurring in about 1–4/10,000 live births. CDH is far more common on the left (75–90%) than the right side (10%) or bilaterally (< 5%). Physiologic reduction of the umbilical hernia at around 12 weeks of gestation is thought to produce sufficient intra-abdominal pressure to cause the viscera to herniate into the thorax [3]. Although significant efforts have been made to unravel the pathophysiology of CDH, the current understanding of the etiology remains inadequate. Recent evidence has suggested that abnormalities linked to the retinoid signaling pathway early in gestation may contribute to the etiology of CDH. This evidence includes: the effect of altering the retinoid system in vitamin A-deficient and transgenic animals; disruption of the retinoid system in teratogen-induced CDH in rodents; the effect of co-administration of retinoids in nitrofen-induced CDH on lung and diaphragm development; and clinical evidence suggesting decreased markers of vitamin A status in human CDH [4].

Right CDH accounts for less than 10% of all diaphragmatic defects and is more difficult to detect than left CDH. The herniated viscera of right CDH consist predominantly of the liver and collapsed bowels. A mediastinal left-shift is a sign strongly suggestive of right CDH. Some sonographic clues can be used to differentiate right CDH from a true lung mass such as a microcystic adenomatoid malformation: the portal vein is within the mass; the gallbladder always herniates with the liver into the thorax; some ascites fluid may be in the chest adjacent to the right lobe of the liver; and the right hypoechoic diaphragmatic muscle may be absent in longitudinal view.

Fetal structural anomalies, regardless of whether they are syndromic or non-syndromic, can be found in 15–45% of CDH cases. Anomalies or syndromes include asplenia/polysplenia, Beckwith-Wiedemann syndrome, and caudal regression syndrome. The prognosis of CDH is greatly influenced by the associated anomalies, degree of prematurity, presence of intrathoracic stomach and liver, lung size, and size of the herniation defect. Survival is poor when associated with anomalies (< 20%) compared with isolated CDH (> 60%) [5].

The mean age at surgery is 3 ± 2 days, the survival rate of uncomplicated CDH is 78%, and there is a low rate of chronic lung disease [6].

The herniated gut always produces a mass effect on the heart and lung. Prenatal diagnosis of small defects or hernias is often difficult. The most important sequelae of CDH are lung hypoplasia and pulmonary hypertension. CDH remains a vexing problem because even though the anatomic defect can be repaired after birth, the pulmonary hypoplasia and vascular physiology might be more problematic [7]. In order to prevent hypoplastic lung before birth, tracheal occlusion has evolved as the mainstream antenatal therapy for this disorder. Accumulation of fluid within the fetal lungs restores lung volume and improves the perinatal outcome in animal studies [8,9]. Nevertheless, Harrison et al reported that tracheal occlusion does not improve human fetal survival [10].

In conclusion, although the prognosis of CDH is still problematic, it seems to be acceptable (> 80%) when the defect is an isolated anomaly and noted during the third trimester, which minimizes the detrimental effects to fetal pulmonary development. The case we presented here had abnormal heart displacement in the thoracic cavity at 34 weeks of gestation, and a lung tumor or diaphragmatic hernia was suspected.

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